# Challenges and Pitfalls in the Evaluation of Structural Variants Callers

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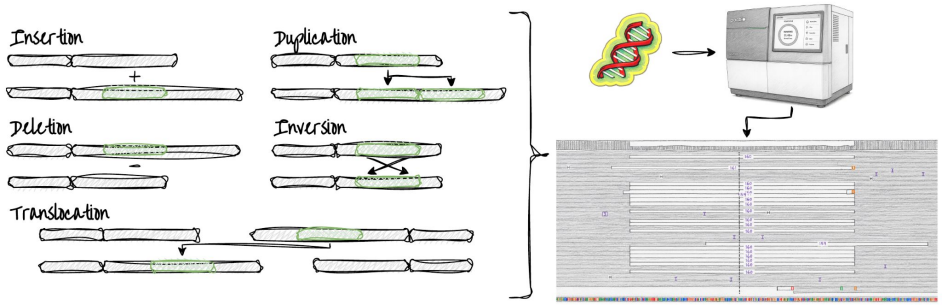
# SV callers evaluation

1. Select reference genome
2. Call SVs from sequencing data
3. Select “ground truth” *(curated or created)*
4. Compare calls
    a. *breakpoint-based + (optional) sequence realignment*
    b. *sequence-based (uncommon)*



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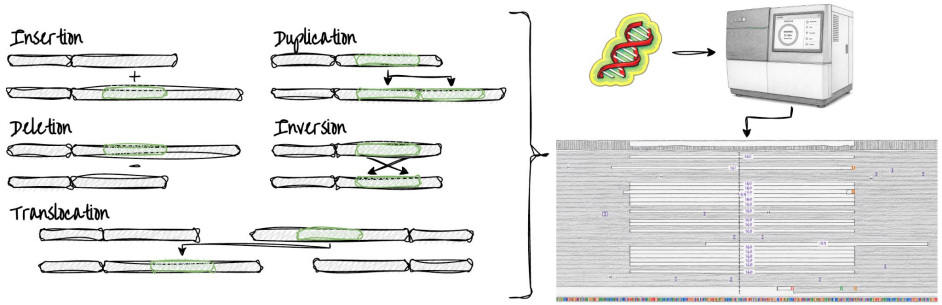
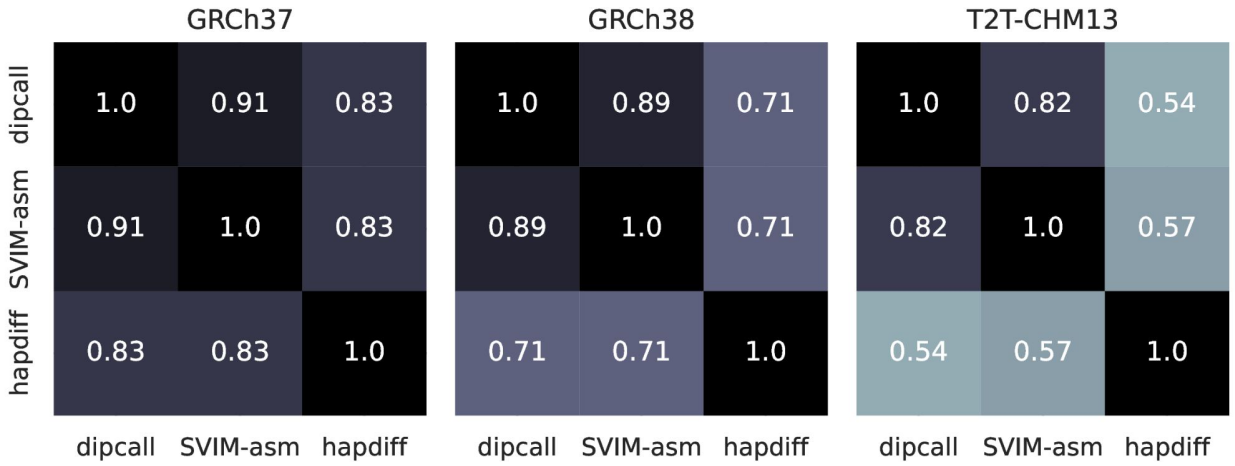



**Inconsistent benchmarking across studies ➪ contradictory findings**

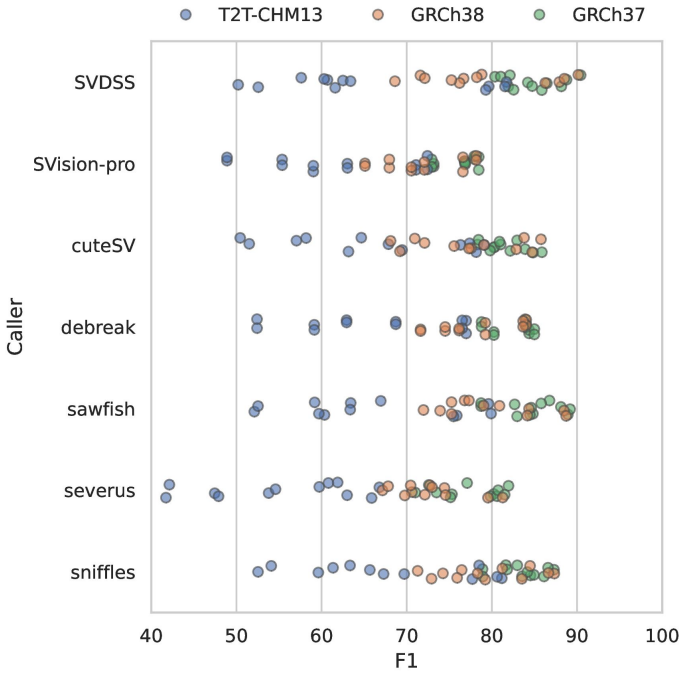
*(our results reveal **expected but overlooked pitfalls** in current evaluation practices)*

# Assembly-based callers




*Leading methods yielded different callsets, even when applied to the same assembly*

# Read-based callers




*Performance depends on ground truth, reference, and benchmark parameters, even when applied to same sample*

# Poster

- *User-defined choices cause overlooked divergence.*
- *Divergence is more evident in complex genomic regions.*
- *Results are not readily transferable across ground truths.*
- *Evaluation against a single ground truth is insufficient.*